



## OXCT1 gene

3-oxoacid CoA-transferase 1

### Normal Function

The *OXCT1* gene provides instruction for making an enzyme called succinyl-CoA:3-ketoacid CoA transferase, often abbreviated as SCOT. The SCOT enzyme is made in the energy-producing centers of cells (mitochondria). The enzyme plays a role in the breakdown of ketones, which are molecules produced by the liver during the breakdown of fats. Ketones are an important source of energy during prolonged periods without food (fasting) or when energy demands are increased, such as during illness or when exercising. In the processing of ketones, the SCOT enzyme converts the molecule acetoacetate to acetoacetyl-CoA.

### Health Conditions Related to Genetic Changes

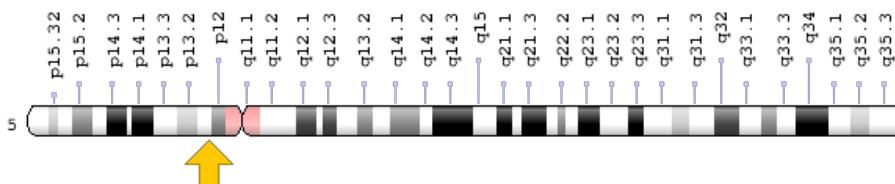
#### succinyl-CoA:3-ketoacid CoA transferase deficiency

At least 20 mutations in the *OXCT1* gene have been found to cause SCOT deficiency, a condition characterized by episodes of extreme tiredness, appetite loss, and seizures, known as ketoacidotic attacks. Most *OXCT1* gene mutations lead to changes in single protein building blocks (amino acids) in the SCOT enzyme and result in an enzyme with little or no function. A reduction in the amount of functional enzyme leads to an inability to break down ketones, often resulting in decreased energy production and an elevated level of ketones in the blood. If these signs become severe, a ketoacidotic attack can occur.

## Chromosomal Location

Cytogenetic Location: 5p13.1, which is the short (p) arm of chromosome 5 at position 13.1

Molecular Location: base pairs 41,730,065 to 41,870,689 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 3-oxoacid CoA transferase 1
- 3-oxoacid-CoA transferase 1
- OXCT
- SCOT
- SCOT1\_HUMAN
- somatic-type succinyl CoA:3-oxoacid CoA-transferase
- somatic-type succinyl-CoA:3-oxoacid-CoA-transferase
- succinyl-CoA:3-ketoacid-CoA transferase
- succinyl-CoA:3-ketoacid-coenzyme A transferase 1, mitochondrial
- succinyl CoA:3-oxoacid CoA transferase

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Ketone Bodies Are a Major Fuel in Some Tissues  
<https://www.ncbi.nlm.nih.gov/books/NBK22387/#A3077>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28OXCT1%5BTIAB%5D%29+OR+%28%28OXCT%5BTIAB%5D%29+OR+%28SCOT%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- 3-OXOACID CoA TRANSFERASE 1  
<http://omim.org/entry/601424>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_OXCT1.html](http://atlasgeneticsoncology.org/Genes/GC_OXCT1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=OXCT1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=8527](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8527)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5019>
- UniProt  
<http://www.uniprot.org/uniprot/P55809>

## **Sources for This Summary**

- OMIM: 3-OXOACID CoA TRANSFERASE 1  
<http://omim.org/entry/601424>
- Berry GT, Fukao T, Mitchell GA, Mazur A, Ciafre M, Gibson J, Kondo N, Palmieri MJ. Neonatal hypoglycaemia in severe succinyl-CoA: 3-oxoacid CoA-transferase deficiency. *J Inherit Metab Dis.* 2001 Oct;24(5):587-95.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/11757586>
- Fukao T, Ishii T, Amano N, Kursula P, Takayanagi M, Murase K, Sakaguchi N, Kondo N, Hasegawa T. A neonatal-onset succinyl-CoA:3-ketoacid CoA transferase (SCOT)-deficient patient with T435N and c.658-666dupAACGTGATT p.N220\_I222dup mutations in the OXCT1 gene. *J Inherit Metab Dis.* 2010 Dec;33 Suppl 3:S307-13. doi: 10.1007/s10545-010-9168-5. Epub 2010 Jul 21.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20652411>

- Fukao T, Mitchell GA, Song XQ, Nakamura H, Kassovska-Bratinova S, Orii KE, Wraith JE, Besley G, Wanders RJ, Niezen-Koning KE, Berry GT, Palmieri M, Kondo N. Succinyl-CoA:3-ketoacid CoA transferase (SCOT): cloning of the human SCOT gene, tertiary structural modeling of the human SCOT monomer, and characterization of three pathogenic mutations. *Genomics*. 2000 Sep 1;68(2):144-51.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/10964512>
- Fukao T, Sass JO, Kursula P, Thimm E, Wendel U, Ficicioglu C, Monastiri K, Guffon N, Baric I, Zabot MT, Kondo N. Clinical and molecular characterization of five patients with succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency. *Biochim Biophys Acta*. 2011 May;1812(5):619-24. doi: 10.1016/j.bbadi.2011.01.015. Epub 2011 Feb 2.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21296660>
- Fukao T, Shintaku H, Kusubae R, Zhang GX, Nakamura K, Kondo M, Kondo N. Patients homozygous for the T435N mutation of succinyl-CoA:3-ketoacid CoA Transferase (SCOT) do not show permanent ketosis. *Pediatr Res*. 2004 Dec;56(6):858-63. Epub 2004 Oct 20.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15496607>

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<https://ghr.nlm.nih.gov/gene/OXCT1>

Reviewed: December 2011

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services